# Maria Isabel Achatz

List of Publications by Year
in descending order

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Version: 2024-02-01

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|---|---|---|---|
| 137 papers | 4,323 citations | 159525 30 h-index | 118793 62 g-index |
| 142 all docs | 142 docs citations | 142 times ranked | 6646 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | TP53 mutations in human cancers: functional selection and impact on cancer prognosis and outcomes. Oncogene, 2007, 26, 2157-2165. | 2.6 | 796 |
| 2 | Cancer Screening Recommendations for Individuals with Li-Fraumeni Syndrome. Clinical Cancer Research, 2017, 23, e38-e45. | 3.2 | 358 |
| 3 | The TP53 mutation, R337H, is associated with Li-Fraumeni and Li-Fraumeni-like syndromes in Brazilian families. Cancer Letters, 2007, 245, 96-102. | 3.2 | 170 |
| 4 | Clinical Management and Tumor Surveillance Recommendations of Inherited Mismatch Repair Deficiency in Childhood. Clinical Cancer Research, 2017, 23, e32-e37. | 3.2 | 157 |
| 5 | Baseline Surveillance in Li-Fraumeni Syndrome Using Whole-Body Magnetic Resonance Imaging. JAMA Oncology, 2017, 3, 1634. | 3.4 | 148 |
| 6 | Revisiting tumor patterns and penetrance in germline TP53 mutation carriers: temporal phases of Liâ€“Fraumeni syndrome. Current Opinion in Oncology, 2018, 30, 23-29. | 1.1 | 129 |
| 7 | PTEN, DICER1, FH, and Their Associated Tumor Susceptibility Syndromes: Clinical Features, Genetics, and Surveillance Recommendations in Childhood. Clinical Cancer Research, 2017, 23, e76-e82. | 3.2 | 128 |
| 8 | Gastric cancer in individuals with Li-Fraumeni syndrome. Genetics in Medicine, 2011, 13, 651-657. | 1.1 | 118 |
| 9 | Detailed haplotype analysis at theTP53locus in p.R337H mutation carriers in the population of Southern Brazil: evidence for a founder effect. Human Mutation, 2010, 31, 143-150. | 1.1 | 116 |
| 10 | Detection of R337H, a germline TP53 mutation predisposing to multiple cancers, in asymptomatic women participating in a breast cancer screening program in Southern Brazil. Cancer Letters, 2008, 261, 21-25. | 3.2 | 94 |
| 11 | Tumor protein 53 mutations and inherited cancer: beyond Li-Fraumeni syndrome. Current Opinion in Oncology, 2010, 22, 64-69. | 1.1 | 91 |
| 12 | Cancer Screening Recommendations and Clinical Management of Inherited Gastrointestinal Cancer Syndromes in Childhood. Clinical Cancer Research, 2017, 23, e107-e114. | 3.2 | 91 |
| 13 | Brain tumors in individuals with familial adenomatous polyposis. Cancer, 2007, 109, 761-766. | 2.0 | 88 |
| 14 | Molecular Characterization of Choroid Plexus Tumors Reveals Novel Clinically Relevant Subgroups. Clinical Cancer Research, 2015, 21, 184-192. | 3.2 | 84 |
| 15 | Germline DNA copy number variation in familial and early-onset breast cancer. Breast Cancer Research, 2012, 14, R24. | 2.2 | 76 |
| 16 | Comprehensive Analysis of BRCA1, BRCA2 and TP53 Germline Mutation and Tumor Characterization: A Portrait of Early-Onset Breast Cancer in Brazil. PLoS ONE, 2013, 8, e57581. | 1.1 | 70 |
| 17 | Highly prevalent TP53 mutation predisposing to many cancers in the Brazilian population: a case for newborn screening?. Lancet Oncology, The, 2009, 10, 920-925. | 5.1 | 67 |
| 18 | Variable population prevalence estimates of germline TP53 variants: A gnomAD-based analysis. Human Mutation, 2019, 40, 97-105. | 1.1 | 66 |

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|---|---|---|---|
| 19 | TP53 PIN3 and MDM2 SNP309 polymorphisms as genetic modifiers in the Li-Fraumeni syndrome: impact on age at first diagnosis. Journal of Medical Genetics, 2009, 46, 766-772. | 1.5 | 64 |
| 20 | The germline mutational landscape of BRCA1 and BRCA2 in Brazil. Scientific Reports, 2018, 8, 9188. | 1.6 | 61 |
| 21 | The Inherited p53 Mutation in the Brazilian Population. Cold Spring Harbor Perspectives in Medicine, 2016, 6, a026195. | 2.9 | 60 |
| 22 | Hereditary breast and ovarian cancer: assessment of point mutations and copy number variations in Brazilian patients. BMC Medical Genetics, 2014, 15, 55. | 2.1 | 57 |
| 23 | Prevalence of the TP53 p.R337H Mutation in Breast Cancer Patients in Brazil. PLoS ONE, 2014, 9, e99893. | 1.1 | 49 |
| 24 | Higher-than-expected population prevalence of potentially pathogenic germlineTP53variants in individuals unselected for cancer history. Human Mutation, 2017, 38, 1723-1730. | 1.1 | 40 |
| 25 | Neoadjuvant Chemotherapy Followed by Interval Debulking Surgery and the Risk of Platinum Resistance in Epithelial Ovarian Cancer. Annals of Surgical Oncology, 2015, 22, 971-978. | 0.7 | 38 |
| 26 | XAF1 as a modifier of p53 function and cancer susceptibility. Science Advances, 2020, 6, eaba3231. | 4.7 | 37 |
| 27 | A Common Molecular Mechanism Underlies Two Phenotypically Distinct 17p13.1 Microdeletion Syndromes. American Journal of Human Genetics, 2010, 87, 631-642. | 2.6 | 36 |
| 28 | Mutational spectrum of the APC and MUTYH genes and genotypeâ€“phenotype correlations in Brazilian FAP, AFAP, and MAP patients. Orphanet Journal of Rare Diseases, 2013, 8, 54. | 1.2 | 35 |
| 29 | TP53 mutation p.R337H in gastric cancer tissues of a 12-year-old male child - evidence for chimerism involving a common mutant founder haplotype: case report. BMC Cancer, 2011, 11, 449. | 1.1 | 34 |
| 30 | Genome-Wide DNA Methylation Analysis Reveals Epigenetic Dysregulation of MicroRNA-34A in TP53-Associated Cancer Susceptibility. Journal of Clinical Oncology, 2016, 34, 3697-3704. | 0.8 | 33 |
| 31 | Characterization of individuals at high risk of developing melanoma in Latin America: bases for genetic counseling in melanoma. Genetics in Medicine, 2016, 18, 727-736. | 1.1 | 31 |
| 32 | Whole-genome sequencing analysis of phenotypic heterogeneity and anticipation in Liâ€“Fraumeni cancer predisposition syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 15497-15501. | 3.3 | 29 |
| 33 | Age at cancer onset in germline TP53 mutation carriers: association with polymorphisms in predicted G-quadruplex structures. Carcinogenesis, 2014, 35, 807-815. | 1.3 | 29 |
| 34 | International distribution and age estimation of the Portuguese BRCA2 c.156_157insAlu founder mutation. Breast Cancer Research and Treatment, 2011, 127, 671-679. | 1.1 | 27 |
| 35 | Li-Fraumeni-like syndrome associated with a large BRCA1 intragenic deletion. BMC Cancer, 2012, 12, 237. | 1.1 | 25 |
| 36 | Recommendations for Advancing the Diagnosis and Management of Hereditary Breast and Ovarian Cancer in Brazil. JCO Global Oncology, 2020, 6, 439-452. | 0.8 | 25 |

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|---|---|---|---|
| 37 | Molecular analysis of TSC1 and TSC2 genes and phenotypic correlations in Brazilian families with tuberous sclerosis. PLoS ONE, 2017, 12, e0185713. | 1.1 | 24 |
| 38 | Mouse Homolog of the Human TP53 R337H Mutation Reveals Its Role in Tumorigenesis. Cancer Research, 2018, 78, 5375-5383. | 0.4 | 24 |
| 39 | Association of TP53 polymorphisms on the risk of Wilms tumor. Pediatric Blood and Cancer, 2014, 61, 436-441. | 0.8 | 23 |
| 40 | Germline <scp>MLH</scp>1, <scp>MSH</scp>2 and <scp>MSH</scp>6 variants in Brazilian patients with colorectal cancer and clinical features suggestive of Lynch Syndrome. Cancer Medicine, 2018, 7, 2078-2088. | 1.3 | 23 |
| 41 | Genomic profiling in ovarian cancer retreated with platinum based chemotherapy presented homologous recombination deficiency and copy number imbalances of CCNE1 and RB1 genes. BMC Cancer, 2019, 19, 422. | 1.1 | 22 |
| 42 | Contribution of rare germline copy number variations and common susceptibility loci in Lynch syndrome patients negative for mutations in the mismatch repair genes. International Journal of Cancer, 2016, 138, 1928-1935. | 2.3 | 21 |
| 43 | Frequency of Thyroid Carcinoma in Brazilian TP53 p.R337H Carriers With Li Fraumeni Syndrome. JAMA Oncology, 2017, 3, 1400. | 3.4 | 21 |
| 44 | Complex Landscape of Germline Variants in Brazilian Patients With Hereditary and Early Onset Breast Cancer. Frontiers in Genetics, 2018, 9, 161. | 1.1 | 21 |
| 45 | Couples coping with screening burden and diagnostic uncertainty in Li-Fraumeni syndrome: Connection versus independence. Journal of Psychosocial Oncology, 2019, 37, 178-193. | 0.6 | 21 |
| 46 | Integration of Genomics in Cancer Care. Journal of Nursing Scholarship, 2013, 45, 43-51. | 1.1 | 20 |
| 47 | LINE-1 hypermethylation in peripheral blood of cutaneous melanoma patients is associated with metastasis. Melanoma Research, 2015, 25, 173-177. | 0.6 | 20 |
| 48 | Rare germline variant (rs78378222) in the TP53 3' UTR: Evidence for a new mechanism of cancer predisposition in Li-Fraumeni syndrome. Cancer Genetics, 2016, 209, 97-106. | 0.2 | 19 |
| 49 | Whole-body magnetic resonance imaging of Li-Fraumeni syndrome patients: observations from a two rounds screening of Brazilian patients. Cancer Imaging, 2018, 18, 27. | 1.2 | 19 |
| 50 | Germline CDKN2A mutations in Brazilian patients of hereditary cutaneous melanoma. Familial Cancer, 2014, 13, 645-649. | 0.9 | 18 |
| 51 | DNA Methylation Levels of Melanoma Risk Genes Are Associated with Clinical Characteristics of Melanoma Patients. BioMed Research International, 2015, 2015, 1-8. | 0.9 | 17 |
| 52 | Comprehensive germline mutation analysis and clinical profile in a large cohort of Brazilian xeroderma pigmentosum patients. Journal of the European Academy of Dermatology and Venereology, 2020, 34, 2392-2401. | 1.3 | 17 |
| 53 | Suggested application of HER2+ breast tumor phenotype for germline TP53 variant classification within ACMG/AMP guidelines. Human Mutation, 2020, 41, 1555-1562. | 1.1 | 16 |
| 54 | Early-onset breast cancer patients in the South and Southeast of Brazil should be tested for the TP53 p.R337H mutation. Genetics and Molecular Biology, 2016, 39, 199-202. | 0.6 | 15 |

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|---|---|---|---|
| 55 | Screening for germline mutations in mismatch repair genes in patients with Lynch syndrome by next generation sequencing. Familial Cancer, 2018, 17, 387-394. | 0.9 | 15 |
| 56 | Prevalence of pathogenic/likely pathogenic variants in the 24 cancer genes of the ACMG Secondary Findings v2.0 list in a large cancer cohort and ethnicity-matched controls. Genome Medicine, 2018, 10, 99. | 3.6 | 15 |
| 57 | Oral and maxillofacial considerations in Gardnerâ€™s syndrome: a report of two cases. Ecancermedicalscience, 2016, 10, 623. | 0.6 | 14 |
| 58 | Germline molecular data in hereditary breast cancer in Brazil: Lessons from a large single-center analysis. PLoS ONE, 2021, 16, e0247363. | 1.1 | 14 |
| 59 | 18F-FDG PET-CT for Surveillance of Brazilian Patients with Li-Fraumeni Syndrome. Frontiers in Oncology, 2015, 5, 38. | 1.3 | 13 |
| 60 | CHEK2 1100DELC germline mutation: a frequency study in hereditary breast and colon cancer Brazilian families. Arquivos De Gastroenterologia, 2012, 49, 273-278. | 0.3 | 12 |
| 61 | The profile and contribution of rare germline copy number variants to cancer risk in Li-Fraumeni patients negative for TP53 mutations. Orphanet Journal of Rare Diseases, 2014, 9, 63. | 1.2 | 12 |
| 62 | The Value of Secondary Cytoreductive Surgery in Recurrent Ovarian Cancer and Application of a Prognostic Score. International Journal of Gynecological Cancer, 2016, 26, 449-55. | 1.2 | 12 |
| 63 | p53 signaling pathway polymorphisms, cancer risk and tumor phenotype in TP53 R337H mutation carriers. Familial Cancer, 2018, 17, 269-274. | 0.9 | 11 |
| 64 | Reproductive factors associated with breast cancer risk in Liâ€“Fraumeni syndrome. European Journal of Cancer, 2019, 116, 199-206. | 1.3 | 10 |
| 65 | Family Health Leaders: Lessons on Living with Liâ€Fraumeni Syndrome across Generations. Family Process, 2020, 59, 1648-1663. | 1.4 | 10 |
| 66 | Number of rare germline CNVs and TP53 mutation types. Orphanet Journal of Rare Diseases, 2012, 7, 101. | 1.2 | 9 |
| 67 | Xeroderma Pigmentosum: Low Prevalence of Germline XPA Mutations in a Brazilian XP Population. International Journal of Molecular Sciences, 2015, 16, 8988-8996. | 1.8 | 9 |
| 68 | Brazilian health-care policy for targeted oncology therapies and companion diagnostic testing. Lancet Oncology, The, 2016, 17, e363-e370. | 5.1 | 9 |
| 69 | ROBO1 deletion as a novel germline alteration in breast and colorectal cancer patients. Tumor Biology, 2016, 37, 3145-3153. | 0.8 | 9 |
| 70 | Haplotype analysis of the internationally distributed BRCA1 c.3331_3334delCAAG founder mutation reveals a common ancestral origin in Iberia. Breast Cancer Research, 2020, 22, 108. | 2.2 | 9 |
| 71 | The breast cancer immunophenotype of TP53-p.R337H carriers is different from that observed among other pathogenic TP53 mutation carriers. Familial Cancer, 2015, 14, 333-336. | 0.9 | 8 |
| 72 | Role of rare germline copy number variation in melanoma-prone patients. Future Oncology, 2016, 12, 1345-1357. | 1.1 | 8 |

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|---|---|---|---|
| 73 | Rare germline alterations in cancer-related genes associated with the risk of multiple primary tumor development. Journal of Molecular Medicine, 2017, 95, 523-533. | 1.7 | 8 |
| 74 | Pheochromocytoma and paraganglioma: implications of germline mutation investigation for treatment, screening, and surveillance. Archives of Endocrinology and Metabolism, 2019, 63, 369-375. | 0.3 | 8 |
| 75 | Ancestry of the Brazilian TP53 c.1010G>A (p.Arg337His, R337H) Founder Mutation: Clues from Haplotyping of Short Tandem Repeats on Chromosome 17p. PLoS ONE, 2015, 10, e0143262. | 1.1 | 8 |
| 76 | Breast Cancer Phenotype Associated With Li-Fraumeni Syndrome: A Brazilian Cohort Enriched by TP53 p.R337H Carriers. Frontiers in Oncology, 2022, 12, 836937. | 1.3 | 8 |
| 77 | Li-Fraumeni Exploration Consortium Data Coordinating Center: Building an Interactive Web-Based Resource for Collaborative International Cancer Epidemiology Research for a Rare Condition. Cancer Epidemiology Biomarkers and Prevention, 2020, 29, 927-935. | 1.1 | 7 |
| 78 | Identification of the TP53 p.R337H Variant in Tumor Genomic Profiling Should Prompt Consideration of Germline Testing for Li-Fraumeni Syndrome. JCO Global Oncology, 2021, 7, 1141-1150. | 0.8 | 7 |
| 79 | A Set of miRNAs, Their Gene and Protein Targets and Stromal Genes Distinguish Early from Late Onset ER Positive Breast Cancer. PLoS ONE, 2016, 11, e0154325. | 1.1 | 7 |
| 80 | HABP2 p.G534E variant in patients with family history of thyroid and breast cancer. Oncotarget, 2017, 8, 40896-40905. | 0.8 | 7 |
| 81 | DNA methylation patterns of candidate genes regulated by thymine DNA glycosylase in patients with TP53 germline mutations. Brazilian Journal of Medical and Biological Research, 2015, 48, 610-615. | 0.7 | 6 |
| 82 | Genome-wide DNA methylation profile of leukocytes from melanoma patients with and without CDKN2A mutations. Experimental and Molecular Pathology, 2014, 97, 425-432. | 0.9 | 5 |
| 83 | Genomic profile of a Li-Fraumeni-like syndrome patient with a 45,X/46,XX karyotype, presenting neither mutations in TP53 nor clinical stigmata of Turner syndrome. Cancer Genetics, 2015, 208, 341-344. | 0.2 | 5 |
| 84 | MIR605 rs2043556 is associated with the occurrence of multiple primary tumors in TP53 p.(Arg337His) mutation carriers. Cancer Genetics, 2020, 240, 54-58. | 0.2 | 5 |
| 85 | Complete Clinical Response in Stage IVB Endometrioid Endometrial Carcinoma after First-Line Pembrolizumab Therapy: Report of a Case with Isolated Loss of PMS2 Protein. Case Reports in Oncology, 2021, 13, 1067-1074. | 0.3 | 5 |
| 86 | Does germ-line deletion of the PIP gene constitute a widespread risk for cancer?. European Journal of Human Genetics, 2014, 22, 307-309. | 1.4 | 4 |
| 87 | Testing Positive on a Multigene Panel Does Not Suffice to Determine Disease Risks. Journal of the National Cancer Institute, 2018, 110, 797-798. | 3.0 | 4 |
| 88 | Germline BAX Deletion in a Patient With Melanoma and Gastrointestinal Stromal Tumor. American Journal of Gastroenterology, 2013, 108, 1372-1375. | 0.2 | 3 |
| 89 | Primary versus interval debulking surgery and the risk to induce platinum resitance.. Journal of Clinical Oncology, 2014, 32, 5588-5588. | 0.8 | 3 |
| 90 | Germline Mutation in MUS81 Resulting in Impaired Protein Stability is Associated with Familial Breast and Thyroid Cancer. Cancers, 2020, 12, 1289. | 1.7 | 3 |

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|---|---|---|---|
| 91 | Genomic Alterations in Patients Showing Multiple Primary Tumors and Family History of Cancer. Annals of Oncology, 2014, 25, iv165. | 0.6 | 2 |
| 92 | Germline large genomic alterations on 7q in patients with multiple primary cancers. Scientific Reports, 2017, 7, 41677. | 1.6 | 2 |
| 93 | Early-Onset Colorectal Cancer in Patients with Li Fraumeni Syndrome: Is It Really Enough to Justify Early Colon Cancer Screening?. Gastroenterology, 2019, 157, 264. | 0.6 | 2 |
| 94 | Prevalence of the Brazilian TP53 Founder c.1010G>A (p.Arg337His) in Lung Adenocarcinoma: Is Genotyping Warranted in All Brazilian Patients?. Frontiers in Genetics, 2021, 12, 606537. | 1.1 | 2 |
| 95 | Keratocystic odontogenic tumor related to nevoid basal cell carcinoma syndrome: clinicopathological study. Brazilian Journal of Oral Sciences, 2013, 12, 23-29. | 0.1 | 2 |
| 96 | Evaluation of rapid whole-body magnetic resonance as screening strategy for early cancer detection in 57 Brazilian Li-Fraumeni syndrome patients.. Journal of Clinical Oncology, 2015, 33, 1534-1534. | 0.8 | 2 |
| 97 | Abstract A37: Complex landscape of germline variants in hereditary and early-onset breast cancer ascertained through whole exome sequencing. , 2018, , . | | 2 |
| 98 | Response to â€œGermline TP53 R337H mutation is not sufficient to establish Li-Fraumeni or Li-Fraumeni-like syndromeâ€ by Ribeiro et al.. Cancer Letters, 2007, 247, 356-358. | 3.2 | 1 |
| 99 | Li-Fraumeni Ontology: A Case Study of an Ontology for Knowledge Discovery in a Cancer Domain. , 2015, , . | | 1 |
| 100 | Response to: Concern regarding classification of germline TP53 variants as likely pathogenic. Human Mutation, 2019, 40, 832-833. | 1.1 | 1 |
| 101 | Utility of interim blood tests for cancer screening in Li-Fraumeni syndrome. Familial Cancer, 2022, 21, 333-336. | 0.9 | 1 |
| 102 | Expanding the phenotype of E318K (c.952Gâ€‰›â€‰A) MITF germline mutation carriers: case series and review of the literature. Hereditary Cancer in Clinical Practice, 2021, 19, 32. | 0.6 | 1 |
| 103 | Molecular Analyses of Early-Onset Gastric Cancer in Brazilian Patients: <i>TP</i>53 Mutations, Cadherin-Catenin and Mucins Proteins Expression. Journal of Cancer Therapy, 2013, 04, 33-42. | 0.1 | 1 |
| 104 | Homologous recombination deficiency and platinum rechallenge in platinum-resistant ovarian cancer patients.. Journal of Clinical Oncology, 2017, 35, 5576-5576. | 0.8 | 1 |
| 105 | Effect of breastfeeding on the risk of breast cancer in Li-Fraumeni syndrome.. Journal of Clinical Oncology, 2018, 36, 1530-1530. | 0.8 | 1 |
| 106 | Abstract P6-08-18: Breast cancer in Li-Fraumeni syndrome and risk-reduction mastectomy inTP53p.R337H carriers. , 2020, , . | | 1 |
| 107 | Novel Insights From the Germline Landscape of Breast Cancer in Brazil. Frontiers in Oncology, 2021, 11, 743231. | 1.3 | 1 |
| 108 | Cancer surveillance for patients with Li-Fraumeni Syndrome in Brazil: A cost-effectiveness analysis. The Lancet Regional Health Americas, 2022, 12, 100265. | 1.5 | 1 |

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|---|---|---|---|
| 109 | 3311 Germline mutations and rare copy number variations in melanoma-prone patients. European Journal of Cancer, 2015, 51, S668. | 1.3 | 0 |
| 110 | Genomic profile of Li-Fraumeni syndrome patients with adrenocortical carcinoma in childhood. Annals of Oncology, 2016, 27, vi11. | 0.6 | 0 |
| 111 | Commentary regarding Schayek et al., entitled â€œThe rate of recurrent BRCA1, BRCA2, and TP53 mutations in the general population, and unselected ovarian cancer cases, in Belo Horizonte, Brazilâ€. Cancer Genetics, 2016, 209, 282-283. | 0.2 | 0 |
| 112 | CN4 Duty to recontact patients previously tested with negative results in a hereditary cancer syndrome center. Annals of Oncology, 2020, 31, S1123-S1124. | 0.6 | 0 |
| 113 | Inherited Pediatric Cancer in Low- and Intermediate-Resource Countries. , 2021, , 361-386. | | 0 |
| 114 | Differential Gene Expression Profiles in Breast Cancer Tumors from Young and Post-Menopausal Patients.. , 2009, , . | | 0 |
| 115 | Abstract 3038: Determination of microRNA expression profile in tumors from young women with breast cancer. , 2010, , . | | 0 |
| 116 | Abstract 1836: Distinct tumor spectrum and age of onset in a Brazilian cohort of p.R337HTP53mutation carriers. , 2011, , . | | 0 |
| 117 | Abstract 5598: Germline TP53 mutation in very early onset breast cancer patients without BRCA1 and BRCA2 mutation in Brazilian population. , 2011, , . | | 0 |
| 118 | Abstract 1837: Clinical diversity and tumor spectrum in Xeroderma Pigmentosum Brazilian patients. , 2011, , . | | 0 |
| 119 | Abstract LB-329: Copy number variations associated with hereditary breast and colorectal carcinomas. , 2011, , . | | 0 |
| 120 | Abstract B10: Germline copy number variation in Li-Fraumeni syndrome patients with TP53 mutations. , 2011, , . | | 0 |
| 121 | Abstract A24: Germline deletion at 3p12.3 in patients with hereditary breast and colorectal carcinoma. , 2011, , . | | 0 |
| 122 | P1-09-07: Contribution of TP53 p.R337H Mutation to Breast Cancer Incidence in Brazil.. , 2011, , . | | 0 |
| 123 | Abstract 5349: Cancer stem cells isolation and characterization from breast tumor of a germline carrier of TP53 p.R337H (Brazilian founder) mutation. , 2012, , . | | 0 |
| 124 | Abstract 3771: Establishment of primary cultures of cancer stem cells from patients with Li-Fraumeni Syndrome, carrier of TP53 p.R337H Brazilian founder mutation.. , 2013, , . | | 0 |
| 125 | Abstract A025: Screening for genomic rearrangements and germline mutations in BRCA1 and BRCA2 genes in hereditary breast cancer unrelated Brazilian families. , 2013, , . | | 0 |
| 126 | Identification of a rare germ-line variant in the TP53 3â€™UTR in individuals with the Li-Fraumeni-like phenotype: A new mechanism of cancer predisposition?. Journal of Clinical Oncology, 2014, 32, 11106-11106. | 0.8 | 0 |

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|---|---|---|---|
| 127 | Abstract 3418: Rare germline copy number variations in hereditary cutaneous melanoma. , 2014, , . | | 0 |
| 128 | Molecular and clinical profile of Li-Fraumeni Syndrome in a Brazilian cohort.. Journal of Clinical Oncology, 2015, 33, e12533-e12533. | 0.8 | 0 |
| 129 | Abstract 2751: Identification of predisposition genes involved in thyroid and breast carcinomas in patients with family history of these tumors by whole exome sequencing. , 2015, , . | | 0 |
| 130 | Abstract 2753: The germline TP53 p.R337H mutation: a putative selective advantage. , 2015, , . | | 0 |
| 131 | Prevalence of germline TP53 p.R337H mutation in Brazilian young breast cancer patients.. Journal of Clinical Oncology, 2017, 35, e13101-e13101. | 0.8 | 0 |
| 132 | Abstract 4287:HABP2p.G534E variant in patients with family history of thyroid and breast cancer. , 2017, , . | | 0 |
| 133 | Abstract 4282: GermlineTP53p.R337H mutations and Li-Fraumeni syndrome: A new variant form of the disease. , 2017, , . | | 0 |
| 134 | Abstract NG05: TP53-mediated human cancer susceptibility is defined by epigenetic dysregulation of microRNA-34A. , 2017, , . | | 0 |
| 135 | Abstract A20: Identification of new promising germline variants in melanoma-prone patients. , 2018, , . | | 0 |
| 136 | Abstract 1247: Hereditary paraganglioma-pheochromocytoma syndrome: Patterns of presentation in a Brazilian oncogenetics clinic. , 2018, , . | | 0 |
| 137 | Abstract 3316: Screening blood tests and cancer detection in Li-Fraumeni syndrome. , 2019, , . | | 0 |